

Editorial: Batten's disease

Batten's disease is a group of rare disorders that present in childhood with retinal and cerebral degeneration and terminate fatally after a prolonged illness. They are inherited as an autosomal recessive condition, with a world wide distribution, and there is as yet no effective treatment. Two cases were described in 1903 by Batten,¹ a physician at the Hospital for Sick Children and the National Hospital for Nervous Diseases, London, though the original description may be attributed to Stengel, who described a Scandinavian family in 1826.² Batten's contribution was to realise that the fundus appearances were different from those of Tay-Sachs's disease, which was described about the same time and later shown to be due to an accumulation of a ganglioside in the brain.³ It soon became apparent that Batten's disease was a group of similar conditions in which autofluorescent material was deposited intracellularly throughout the body and that the various subtypes of the condition could be recognised from its ultrastructural appearances. There is at present no agreement on the composition of this substance, though Zeman and Dyken⁴ postulated that the material was due to the accumulation of ceroid and lipofuscin and suggested Batten's disease should be called neuronal ceroid lipofuscinosis, but recently it has been suggested that vitamin A metabolism might be implicated in the late infantile form.

In this issue of the *BJO* Spalton and colleagues review juvenile Batten's disease. This is the subtype of the disease of most interest to ophthalmologists, since these children present with visual failure. There are about a dozen new cases of Batten's disease a year in Britain, and the juvenile type accounts for a quarter of these. To put this in perspective, there are approximately 100 new blind registrations a year in England and Wales in the 5-15 age group, and since most of these registrations are due to congenital anomalies acquired forms of blindness are unusual. Batten's disease must constitute a significant proportion of children with acquired retinal blindness. Most ophthalmologists will see only an occasional case in the course of a professional career, but early diagnosis of the disease is nonetheless important so that parents can receive genetic counselling and medical advice. There is a 1:4 risk of further children being affected, and there is no method of prenatal diagnosis at present.

The rarity of neurodegenerative diseases presents

a considerable diagnostic challenge and the ophthalmologist should appreciate the significant diagnostic contribution he can make. Careful fundus examination is usually necessary for recognition of the cherry red spot which occurs in Tay-Sachs disease (infantile GM₁, GM₂ types 1 and 2), sialidase deficiency (type 1+2), and acute neuronopathic Niemann-Pick disease (type A). Pigmentary retinopathy may be rarely associated with abeta lipoproteinaemia or Refsum's disease, and recognition of these conditions may have therapeutic implications. Visual loss may also be due to cortical blindness, and adrenoleucodystrophy typically presents in this way. Ocular motor abnormalities may also provide clues, for a vertical supranuclear ophthalmoplegia is often a helpful diagnostic sign of the ophthalmoplegic lipidosis syndrome (with sea blue histocytes), and a horizontal supranuclear ophthalmoplegia may be seen in late onset Gaucher's disease.

Children with visual problems usually have severe visual loss before they come to the ophthalmologist. Some of them will have endocrine or neurological symptoms suggesting a systemic process, but those patients without other signs are the most difficult to deal with. The ophthalmologist must consider compressive lesions (craniopharyngiomas, gliomas), inflammatory conditions (subacute sclerosing panencephalitis), or degenerative disorders. This group with organic disease can usually be separated from children with hysterical blindness by performing a visual evoked response in an experienced department (though hysterical symptoms may mask an underlying organic illness). The variety of further investigations available include neurophysiological procedures, endocrine assays, and computed tomography, and there may be advantages in staff with a special experience with children undertaking them.

References

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- 2 Stengel E. Beretning om et maerkeligt Sygdomstilfoelde hos fire sodskende; Naenheden af Roraas. *Eyr et Medicinsk Tidsskrift* 1826; **1**: 347-52.
- 3 Klenk E. Beitrage zur cherrie der lipidosen, Niemann-Picksche Krankheit und amaurotische Idiotie. *Hoppe Seylers Z Physiol Chem* 1939; **262**: 128-43.
- 4 Zeman W, Dyken P. Neuronal ceroid-lipofuscinosis (Batten's disease) relationship to amaurotic family idiocy. *Pediatrics* 1969; **44**: 570-83.